British Society for Genetic Medicine Annual Conference
- UK Clinical Genomics 2018 -

Tuesday 2nd October 2018 | Royal College of Physicians | Regent's Park | London | NW1 4LE

09:00 – 10.00	**Registration and Coffee**	
09:55 – 10:00	**Welcome**	**Anneke Lucassen** *(Chair, BSGM)*
10:00 – 12:10	**UK Clinical Genomics in practice update (Wolfson Theatre)** *Chairs:* ***Dominic McMullan and Carol Gardiner***	
10:00 – 10:40	Insights into Developmental Disorders from the DDD and PAGE studies	**Matt Hurles** *(Welcome Sanger Institute)*
10:40 – 11:00	Ethical and practical issues of PAGE	**Ruth Horn** *(University of Oxford)*
11:00 – 11:20	Avoid unsolicited findings or findings that cannot be interpreted in genome sequencing	**Martina Cornel** *(VU University Medical Center, Amsterdam)*
11:20 – 11:30	Discussion and Panel Q & A The ethical aspects of modern genomic technologies	**Anneke Lucassen** *(BSGM Chair)* **and Alison Hall** *(PGH Foundation)*
11:30 – 11:50	Have European Reference Networks a part to play in the Genomic Era?	**Jill Clayton-Smith** *(University of Manchester)*
11:50 – 12:10	A systematic framework for the interpretation of copy number variants	**Erin Rooney Riggs** *(ClinGen Consortium)*
	Lunch and Trade Exhibition 12:15 – 13:05 NIHR Early Career Researcher Meeting: How to get started in research (Dorchester Library)	
13:10 – 14:40	**Concurrent submitted abstract sessions**	
	Discovery - Wolfson Theatre *Chairs:* ***Diana Baralle*** *and* ***Emma Woodward***	**Service - Dorchester Library** Chairs: **Mohnish Suri** and **Sarah Gibson**
13.10	Clinical utility of mutational signatures derived from whole genome sequencing in breast cancer. **Helen Davies.**	Reduced penetrance X chromosome aneuploidy in a population-based cohort of >244,000 females from the UK Biobank. **Anna Murray.**
13.25	The Next Generation Children (NGC) project: A paradigm-shifting model for translational genomics with rapid integration into healthcare. **Isabelle Delon.**	The surprising lifestyle intervention that facilitates brain neuroplasticity in Bardet-Biedl syndrome. **Elizabeth Forsythe.**
13.40	Non-invasive prenatal diagnosis (NIPD) of single gene disorders (SGDs): 18 months of relative haplotype dosage (RHDO) analysis. **Stephanie K Allen.**	Mainstreaming BRCA mutation testing for ovarian cancer patients: addressing health inequalities. **Alice Marsden.**
13.55	Classification of 5' non-coding variants of BRCA1 by saturation genome editing. **Elke van Veen.**	FFPE testing in deceased family members: implications for clinical management of patients seen in the genetics clinic. **Harry Fraser.**
14.10	Utilisation of whole genome sequences for extraction of pharmacogenomic data. **Joanna Kenny.**	A recurrent de novo duplication in the ATAD3 gene cluster causes a severe congenital neurodevelopmental disorder. **Adam Gunning.**
14.25	Clinically-relevant postzygotic mosaicism in parents and children identified from trio exome sequencing data. **Caroline Wright.**	The PREGCARE study – Personalised, pre-conception recurrence risk stratification of families with a child affected by a disorder caused by a de novo mutation. **Marie Bernkopf.**
	Afternoon Tea and Trade Exhibition	
15:10 – 17:30	**Applications of Genomics in Management of Human Disease (Wolfson Theatre)** *Chairs:* **Anna Middleton** and **Marc Tischowitz**	
15:10 – 15:40	Meeting the therapeutic challenge of Huntington's disease	**Sarah Tabrizi** *(University College London)*
15:40 – 16:10	Genome editing and human reproduction	**Andy Greenfield** *(MRC Harwell Institute, Oxford)*
16:10 – 16:40	Pharmacogenomics of G-protein coupled receptor drug targets	**Madan Babu** *(MRC Laboratory of Molecular Biology, Cambridge)*
16:40 – 17:30	**The BSGM Lecture 2018** : Deciphering the function of the non-coding genome	**Wendy Bickmore** *(MRC Human Genetics Unit, University of Edinburgh)*
17:30 – 18:00	**BSGM Annual General Meeting**	